Single Nucleotide Polymorphism

NCBI

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Reference SNP Cluster Report

NCBI SNP CLUSTER ID:	**rs7592**
Organism:	human (*Homo sapiens*)
Variation Class:	SNP: single nucleotide polymorphism
Molecule Type:	Genomic
dbSNP build of first appearance:	52
dbSNP build of most recent change to cluster:	116

SNP Details are categorized in the following sections:
Submission   Fasta   Resource   Locus   Map   Variation   Va

Submitter records for this RefSNP Cluster

The submission **ss11944722** has the longest flanking sequence of all cluster members and was us current build.

NCBI Assay ID	Handle|Submitter ID	Validation Status	Entry Date	Update Date
ss9489	CGAP-GAI|50173	✗	08/23/99	01/29/0
ss20257	UWGC|2197		11/15/99	01/29/0
ss20261	UWGC|2201		11/15/99	01/29/0
ss1510338	LEE|34488		09/13/00	01/04/0
ss1510475	LEE|38702		09/13/00	01/04/0
ss3194964	YUSUKE|IMS-JST010882		08/07/01	01/04/0
ss4404192	LEE|e34488		04/26/02	07/03/0
ss4404281	LEE|e38702		04/26/02	07/03/0
ss11944722	WI_SSAHASNP|chr8.NT_023666.15_396196		07/04/03	07/04/0

Fasta sequence   ([Legend](#))

```
>gnl|dbSNP|rs7592|allelePos=201|totalLen=401|taxid=9606|snpclass=1|alleles='A/G'|mol=Genom

TCATCCAGGC AACTCGGGGG AGGGGAAGCT CACAGACCGG TACTTCCCAC TCCCCTGATT
CTCTCTGTCC ATCCTCAACA TTCCTTTGCT TCACAGGGTC AGTGGAAGCC CCAACGGGAA
AGGAAACGCC CCGGGCAAAG GGTCTTTTGC AGCTTTTGCA GACGGGCAAG AAGCTGCTTC
TGCCCACACC GCAGGGACAA
R
```

```
CCCTGGAGAA ATGGGAGCTT GGGGAGAGGA TGGGAGTGGG CAGAGGTGGC GCCCAGGGGC
CCGGGAACTC CTGCCACAAC AGAATAAAGC AGCCTGATTG AAAAGCAAAG GGTCTGCTTC
TGTCTTCCTG CAGGGCGCAG TCCTGCCCTG GCGGGGGCCG GCCAAGAAGG GAAGGGCCTT
GGGAGAGCAA AGTGGGGTTT
```

NCBI Resource Links

Submitter-Referenced Accessions:
dbSTS:
GenBank: U02948.1 Hs.1074 NM_003018

dbSNP Blast Analysis:
NCBI RefSeq NM (mRNA): NM_003018.1
GenBank HTGS Finished: AC105141.7 AC105206.8 J03890.1 U02948.1
GenBank STS: G62003.1 G62006.1
GenBank mRNA: AK058094.1 BC005913.1 J03517.1 J03553.1

UniGene transcribed sequence cluster:
UniGene Cluster ID: 1074

LocusLink Analysis

LocusLink via analysis of contig annotation: BMP1 bone morphogenetic protein 1

Variation has been mapped within 2 kb of a mRNA transcript for bone morphogenetic protein 1

Contig accession	Contig position	Protein accession	Function	dbSNP allele	Protein residue	Codon position	Amino acid position
NT_023666	396196		locus				

LocusLink via analysis of contig annotation: SFTPC surfactant, pulmonary-associated protein C

Gene Model (contig mRNA transcript) information from genome sequence for NM_003018

Contig accession	Contig position	Protein accession	Function	dbSNP allele	Protein residue	Codon position	Amino acid position
NT_023666	396196		untranslated region				

LocusLink via BLAST analysis of mRNAs: SFTPC surfactant, pulmonary-associated protein C
Variations are assigned to a gene if mapped within 2 kb of mRNA sequence feature.

Accession class	Nucleotide accession	Nucleotide Position	Hit orientation	Protein accession	Function
NCBI RefSeq	NM_003018.1	744	plus strand	NP_003009.1	unclassified
GenBank mRNA	BC005913.1	740	plus strand	AAH05913.1	unclassified
GenBank mRNA	J03517.1	744	plus strand	AAA36634.1	unclassified
GenBank mRNA	J03553.1	891	plus strand	AAA36631.1	unclassified

Integrated Maps:

NCBI MapViewer: rs7592 maps exactly once on NCBI human chromosome 8

Chromosome	Contig accession	Contig Position	Chromosome Position	Hit orientation

8   NT_023666.15 396196   21842452   plus strand

NCBI Sequence Viewer: See rs7592 in Sequence Viewer.

Project Ensembl: Query rs7592 in Ensembl.

UC Santa Cruz Genome Assembly: Query rs7592 on the Santa Cruz Assembly.

Variation Summary:

Assay sample size (number of chromosomes):   101
Population data sample size (number of chromosomes): 188
Total number of populations with frequency data: 1
Total number of individuals with genotype data: 0
Average estimated heterozygosity: 0.488
Average Allele Frequency:
          A   0.576
          G   0.424

Validation Summary:

Validation status:

Marker displays Mendelian segregation: UNKNOWN
PCR results confirmed in multiple reactions: UNKNOWN
Homozygotes detected in individual genotype data: UNKNOWN

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|New Batches | Method | Population | Publication
| Chromosome Report | Batch |Locus Info | Free Form | Easy Form | Between
Marker
HAPLOTYPE: Specifications | Sample HapSet | Sample Individual
NCBI: PubMed | Entrez | BLAST | OMIM | Taxomomy | Structure

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Locus Information
STS Markers
Free Form Search
  Simple
  Advanced

HAPLOTYPE
Specifications
Sample HapSet
Sample Individual

Reference SNP Cluster Report

NCBI SNP CLUSTER ID:	rs1139547
Organism:	human (*Homo sapiens*)
Variation Class:	SNP: single nucleotide polymorphism
Molecule Type:	cDNA
dbSNP build of first appearance:	86
dbSNP build of most recent change to cluster:	103

SNP Details are categorized in the following sections:
Submission   Fasta   Resource   Locus   Map   Variation   Va

Submitter records for this RefSNP Cluster

The submission **ss4322597** has the longest flanking sequence of all cluster members and was use during BLAST analysis for the current build.

NCBI Assay ID	Handle|Submitter ID	Validation Status	Entry Date	Update Date	Build Added	Molecule Type	Sequen Ori ntat
ss1548788	LEE|1015779		09/13/00	01/29/01	86	cDNA	forward
ss4322597	CGAP-GAI|263851		11/28/01	03/26/02	103	cDNA	reverse

Fasta sequence   (Legend)

```
>gnl|dbSNP|rs1139547|allelePos=51|totalLen=107|taxid=9606|snpclass=1|alleles='A/T/G'|mol=cDN

CCCTGGAG
AAATGGGAGC  TTGGGGAGAG  GATGGGAGTG  GGCAGAGGTG  GC
D
CCCAGGGGCC  CGGGAACTCC  TGCCACAACA  GAATAAAGCA  GCCTGATTGA  AAAGCA
```

NCBI Resource Links

Submitter-Referenced Accessions:
dbSTS:
GenBank: AI692500 Hs.1074

dbSNP Blast Analysis:
NCBI RefSeq NM (mRNA): NM_003018.1
GenBank STS: G62003.1 G62006.1
GenBank mRNA: AK058094.1 BC005913.1 J03517.1 J03553.1

LocusLink Analysis

LocusLink via analysis of contig annotation: BMP1 bone morphogenetic protein 1

Variation has been mapped within 2 kb of a mRNA transcript for bone morphogenetic protein 1

Contig accession	Contig position	Protein accession	Function	dbSNP allele	Protein residue	Codon position	Amino acid position
NT_023666	396247		locus				

LocusLink via analysis of contig annotation: SFTPC surfactant, pulmonary-associated protein C

Gene Model (contig mRNA transcript) information from genome sequence for NM_003018

Contig accession	Contig position	Protein accession	Function	dbSNP allele	Protein residue	Codon position	Amino acid position
NT_023666	396247		untranslated region				

LocusLink: no link established by BLAST analysis of mRNA sequences

Integrated Maps:

NCBI MapViewer: rs1139547 maps exactly once on NCBI human chromosome 8

Chromosome	Contig accession	Contig Position	Chromosome Position	Hit orientation
8	NT_023666.15	396247	21842503	plus strand

NCBI Sequence Viewer: See rs1139547 in Sequence Viewer.

Project Ensembl: Query rs1139547 in Ensembl.

UC Santa Cruz Genome Assembly: Query rs1139547 on the Santa Cruz Assembly.

Variation Summary:

Assay sample size (number of chromosomes):	31
Population data sample size (number of chromosomes):	
Total number of populations with frequency data:	0
Total number of individuals with genotype data:	0
Average estimated heterozygosity:	Not available

Validation Summary:

Validation status:

Marker displays Mendelian segregation:	UNKNOWN
PCR results confirmed in multiple reactions:	UNKNOWN
Homozygotes detected in individual genotype data:	UNKNOWN

GENERAL: Contact Us | Homepage | Announcements |dbSNP Summary | Genome | FTP SERVER | Build History | Handle Request

DOCUMENTATION: FAQ | Overview | How to Submit | RefSNP Summary
Info | Database Schema
SEARCH: Entrez SNP | Blast SNP | Main Search |Batch Query | By Submitter
|New Batches | Method | Population | Publication
| Chromosome Report | Batch |Locus Info | Free Form | Easy Form | Between
Marker
HAPLOTYPE: Specifications | Sample HapSet | Sample Individual
NCBI: PubMed | Entrez | BLAST | OMIM | Taxomomy | Structure

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Reference SNP Cluster Report

NCBI SNP CLUSTER ID:	**rs1126931**
Organism:	human (*Homo sapiens*)
Variation Class:	SNP: single nucleotide polymorphism
Molecule Type:	cDNA
dbSNP build of first appearance:	86
dbSNP build of most recent change to cluster:	103

SNP Details are categorized in the following sections:
Submission   Fasta   Resource   Locus   Map   Variation   Va

Submitter records for this RefSNP Cluster

The submission **ss1510417** has the longest flanking sequence of all cluster members and was use during BLAST analysis for the current build.

NCBI Assay ID	Handle|Submitter ID	Validation Status	Entry Date	Update Date	Build Added	Molecule Type	Sequen Ori ntat
ss1510417	LEE|36582		09/13/00	01/29/01	86	cDNA	forward
ss4322596	CGAP-GAI|263848		11/28/01	03/26/02	103	cDNA	reverse

Fasta sequence   (Legend)

```
>gnl|dbSNP|rs1126931|allelePos=51|totalLen=101|taxid=9606|snpclass=1|alleles='C/T'|mol=cDNA

ACCCTGGAGA AATGGGAGCT TGGGGAGAGG ATGGGAGTGG GCAGAGGTGG
Y
ACCCAGGGGC CCGGGAACTC CTGCCACAAC AGAATAAAGC AGCCTGATTG
```

NCBI Resource Links

Submitter-Referenced Accessions:
dbSTS:
GenBank: AI203517 Hs.1074

dbSNP Blast Analysis:
NCBI RefSeq NM (mRNA): NM_003018.1
GenBank HTGS Finished: AC105141.7 AC105206.8 J03890.1 U02948.1
GenBank STS: G62003.1 G62006.1
GenBank mRNA: AK058094.1 BC005913.1 J03517.1 J03553.1

UniGene transcribed sequence cluster:

UniGene Cluster ID: 1074

LocusLink Analysis

LocusLink via analysis of contig annotation: BMP1 bone morphogenetic protein 1

Variation has been mapped within 2 kb of a mRNA transcript for bone morphogenetic protein 1

Contig accession	Contig position	Protein accession	Function	dbSNP allele	Protein residue	Codon position	Amino acid position
NT_023666	396246		locus				

LocusLink via analysis of contig annotation: SFTPC surfactant, pulmonary-associated protein C

Gene Model (contig mRNA transcript) information from genome sequence for NM_003018

Contig accession	Contig position	Protein accession	Function	dbSNP allele	Protein residue	Codon position	Amino acid position
NT_023666	396246		untranslated region				

LocusLink via BLAST analysis of mRNAs: SFTPC surfactant, pulmonary-associated protein C
Variations are assigned to a gene if mapped within 2 kb of mRNA sequence feature.

Accession class	Nucleotide accession	Nucleotide Position	Hit orientation	Protein accession	Function
NCBI RefSeq	NM_003018.1	794	plus strand	NP_003009.1	unclassified
GenBank mRNA	BC005913.1	790	plus strand	AAH05913.1	unclassified
GenBank mRNA	J03517.1	794	plus strand	AAA36634.1	unclassified
GenBank mRNA	J03553.1	941	plus strand	AAA36631.1	unclassified

Integrated Maps:

NCBI MapViewer: rs1126931 maps exactly once on NCBI human chromosome 8

Chromosome	Contig accession	Contig Position	Chromosome Position	Hit orientation
8	NT_023666.15	396246	21842502	plus strand

NCBI Sequence Viewer: See rs1126931 in Sequence Viewer.

Project Ensembl: Query rs1126931 in Ensembl.

UC Santa Cruz Genome Assembly: Query rs1126931 on the Santa Cruz Assembly.

Variation Summary:

Assay sample size (number of chromosomes):	36
Population data sample size (number of chromosomes):	
Total number of populations with frequency data:	0
Total number of individuals with genotype data:	0
Average estimated heterozygosity:	Not available

Validation Summary:

Validation status:

Marker displays Mendelian segregation: UNKNOWN
PCR results confirmed in multiple reactions: UNKNOWN
Homozygotes detected in individual genotype data: UNKNOWN

GENERAL: Contact Us | Homepage | Announcements |dbSNP Summary |
Genome | FTP SERVER | Build History | Handle Request
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Marker
HAPLOTYPE: Specifications | Sample HapSet | Sample Individual
NCBI: PubMed | Entrez | BLAST | OMIM | Taxomomy | Structure

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```
        FILE 'MEDLINE' ENTERED AT 14:17:35 ON 10 SEP 2003
                ACTIVATE SURFACTANT/L
                ---------
L1  (     1623)SEA PLU=ON   SFTPC OR SFTP2 OR SP-C OR SP5 OR SPC OR (SFTP(W)(C
              OR 2)) OR (SURFACTANT(3A)PULMONARY(3A)ASSOCIATED(3A)(C OR 2))
              OR (PULMONARY(3A)SURFACTANT(3A)APOPROTEIN(5A)(2 OR C)) OR
              (SURFACTANT(3A)PROTEOLIPID) OR (PULMONARY SURFACTANT PROTEIN
              SP5)
L2  (     2417)SEA PLU=ON   SFTPC OR SFTP2 OR SP-C OR SP5 OR SPC OR (SFTP(W)(C
              OR 2)) OR (SURFACTANT(3A)PULMONARY(3A)ASSOCIATED(3A)(C OR 2))
              OR (PULMONARY(3A)SURFACTANT(3A)APOPROTEIN(5A)(2 OR C)) OR
              (SURFACTANT(3A)PROTEOLIPID) OR (PULMONARY SURFACTANT PROTEIN
              SP5)
L3  (     4226)SEA PLU=ON   SFTPC OR SFTP2 OR SP-C OR SP5 OR SPC OR (SFTP(W)(C
              OR 2)) OR (SURFACTANT(3A)PULMONARY(3A)ASSOCIATED(3A)(C OR 2))
              OR (PULMONARY(3A)SURFACTANT(3A)APOPROTEIN(5A)(2 OR C)) OR
              (SURFACTANT(3A)PROTEOLIPID) OR (PULMONARY SURFACTANT PROTEIN
              SP5)
L4  (     8266)SEA PLU=ON   SFTPC OR SFTP2 OR SP-C OR SP5 OR SPC OR (SFTP(W)(C
              OR 2)) OR (SURFACTANT(3A) PULMONARY(3A) ASSOCIATED(3A)(C OR
              2)) OR (PULMONARY(3A) SURFACTANT(3A) APOPROTEIN(5A)(2 OR C))
              OR (SURFACTANT(3A) PROTEOLIPID) OR (PULMONARY SURFACTANT
              PROTEIN SP5)
L5  (      603)SEA PLU=ON   L1 AND SURFACTANT
L6  (      722)SEA PLU=ON   L2 AND SURFACTANT
L7  (      846)SEA PLU=ON   L3 AND SURFACTANT
L8  (     2171)SEA PLU=ON   L4 AND SURFACTANT
L9  (       18)SEA PLU=ON   L5 AND (L1(8A)(SNP OR POLYMORPH? OR ALLEL? OR
              MUTAT? OR VARIAN? OR RFLP OR RESTRICTION))
L10 (       26)SEA PLU=ON   L6 AND (L2(8A)(SNP OR POLYMORPH? OR ALLEL? OR
              MUTAT? OR VARIAN? OR RFLP OR RESTRICTION))
L11 (       18)SEA PLU=ON   L7 AND (L3(8A)(SNP OR POLYMORPH? OR ALLEL? OR
              MUTAT? OR VARIAN? OR RFLP OR RESTRICTION))
L12 (       62)SEA PLU=ON   L8 AND (L4(8A)(SNP OR POLYMORPH? OR ALLEL? OR
              MUTAT? OR VARIAN? OR RFLP OR RESTRICTION))
L13 (       37)DUP REM L12 (25 DUPLICATES REMOVED)
                ---------

        FILE 'MEDLINE, BIOSIS, CAPLUS' ENTERED AT 14:18:25 ON 10 SEP 2003
L14        44 SEA PLU=ON   L12
L15      8265 SEA PLU=ON   SFTPC OR SFTP2 OR SP-C OR SP5 OR SPC OR (SFTP(W)(C
              OR 2)) OR (SURFACTANT(3A) PULMONARY(3A) ASSOCIATED(3A)(C OR
              2)) OR (PULMONARY(3A) SURFACTANT(3A) APOPROTEIN(5A)(2 OR C))
              OR (SURFACTANT(3A) PROTEOLIPID) OR (PULMONARY SURFACTANT
              PROTEIN SP5)
L16      2171 SEA PLU=ON   L15 AND SURFACTANT
L17        62 SEA PLU=ON   L16 AND (L1(8A)(SNP OR POLYMORPH? OR ALLEL? OR
              MUTAT? OR VARIAN? OR RFLP OR RESTRICTION))
L18        37 DUP REM L17 (25 DUPLICATES REMOVED)
              D TI 1-37
              D IBIB AB 35
              D KWIC 35
              D IBIB AB 27
              D IBIB AB 2-4 8 11 14 15 17 18 20
        FILE 'MEDLINE, BIOSIS, CAPLUS' ENTERED AT 15:00:57 ON 10 SEP 2003
L22        24 SEA PLU=ON   NOGEE-L/AU
              E NOGEE-L/AU
              E NOGEE L/AU
L23       127 SEA PLU=ON   ("NOGEE L"/AU OR "NOGEE L M"/AU OR "NOGEE LARRY"/AU
              OR "NOGEE LAWRENCE"/AU OR "NOGEE LAWRENCE M"/AU)
              E COLE F/AU
              E WHITSETT J/AU
```

```
L24          0 SEA PLU=ON   "WHITSETT J"/AU "WHITSETT J A"/AU
               E WHITSETT J/AU
L25        647 SEA PLU=ON   ("WHITSETT J"/AU OR "WHITSETT J A"/AU)
               E WHITSETT JE/AU
L26        597 SEA PLU=ON   ("WHITSETT JE"/AU OR "WHITSETT JEFF"/AU OR
               "WHITSETT JEFF A"/AU OR "WHITSETT JEFFERY A"/AU OR "WHITSETT
               JEFFFREY A"/AU OR "WHITSETT JEFFREY"/AU OR "WHITSETT JEFFREY
               A"/AU)
L27       1324 SEA PLU=ON   L26 OR L25 OR L23
L28        987 SEA PLU=ON   L27 AND SURFACTANT
               D KWIC 1-5
L29        320 SEA PLU=ON   L28 AND L16
L30        305 SEA PLU=ON   L29 NOT L18
L31        303 SEA PLU=ON   L29 NOT L17
L32         13 SEA PLU=ON   L31 AND INTERSTITIAL
L33          7 DUP REM L32 (6 DUPLICATES REMOVED)
               D TI 1-7
               D IBIB AB 3
```

```
                    GenCore version 5.1.6
              Copyright (c) 1993 - 2003  Compugen Ltd.


OM nucleic - nucleic search, using sw model

Run on:          September 16, 2003, 13:07:51 ; Search time 2680 Seconds
                                        (without alignments)
                                        351.090 Million cell updates/sec


Title:           US-10-074-247-1-C-AT-243
Perfect score:   23
Sequence:        1 ggagatgagcactggggcgccgg 23


Scoring table:   OLIGO_NUC
                 Gapop 60.0 , Gapext 60.0


Searched:        2888711 seqs, 20454813386 residues


Word size :      12


Total number of hits satisfying chosen parameters:      21885


Minimum DB seq length: 0
Maximum DB seq length: 2000000000


Post-processing: Listing first 500 summaries


Database :       GenEmbl:*
identical search repeated in GENSEQ, USPATS, USPGPUBS, and EST databases
```